TAY-SACHS DISEASE

BASIC INFORMATION

DESCRIPTION
An inherited, rare disorder of the central nervous system in infants and young children. It causes progressive impairment and early death. Less than 100 children are born with the disease each year in the U.S.

FREQUENT SIGNS AND SYMPTOMS
The child seems normal at birth. Between 3 and 6 months, the following symptoms begin to appear:
- Loss of alertness and retarded mental development.
- Loss of muscle strength, such as difficulty sitting up or turning over.
- Deafness.
- Blindness.
- Severe constipation caused by an impaired nerve supply to the colon.
- Seizures.

CAUSES
An inherited disease resulting from a recessive gene that causes enzyme deficiency. If both parents have the gene, they have a 25% chance of having a child with Tay-Sachs disease. If only one parent is a carrier, the children will not have the disease. The gene occurs in 1 out of 60 people of Ashkenazi Jewish or French Canadian ancestry.

RISK INCREASES WITH
Genetic factors. Most parents who carry the recessive gene are of Eastern European Jewish (Ashkenazi) or French Canadian origin .

PREVENTIVE MEASURES
- Obtain genetic screening for children in families with Tay-Sachs.
- Obtain genetic counseling if you or your spouse have a family history of Tay-Sachs or are of Ashkenazi or French Canadian background.
- If you are expecting a child and have a family history of Tay-Sachs, consider amniocentesis to detect if the fetus has the disease.

EXPECTED OUTCOMES
Death usually occurs before age 5.

POSSIBLE COMPLICATIONS
- Pneumonia.
- Pressure sores.

TREATMENT

GENERAL MEASURES
- Time in an extended-care facility for basic care if parents are unable to provide it at home.
- Psychotherapy or counseling for parents and siblings to learn to cope with the distress produced by this condition.
- Seek out support groups for families of Tay-Sachs victims.
- Additional information available from the National Tay-Sachs and Allied Disease Association, 2001 Beacon St., Suite 204, Brookline, MA 02146, (617) 277-4463.

MEDICATIONS
- Anticonvulsants to control seizures.
- Stool softeners and laxatives to relieve constipation.
- Other medicines to control complicating disorders as they arise.

ACTIVITY
In the early stages, encourage the child to be as active as possible. Increasing mental, nervous and muscular deficiencies will eventually confine the child to bed much of the time.

DIET
Provide adequate fluids and a normal, high-fiber diet to minimize constipation. Feeding by tube usually becomes necessary as the disease progresses.

NOTIFY OUR OFFICE IF
- You are concerned about your infant's mental and physical development.
- You think you or any member of your family carries the abnormal gene. A genetic counselor can advise you on how to prevent having children with this disease.

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